

WHAT IS CLAIMED IS:

1. An antibody that binds to a polypeptide comprising an amino acid sequence as set forth in SEQ ID NO: 2 or SEQ ID NO: 4, or a fragment of said polypeptide.

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2. The antibody of claim 1, wherein the polypeptide is from a mammal.

3. The antibody of claim 2, wherein the polypeptide is from a human.

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4. The antibody of claim 1, wherein the antibody is monoclonal.

5. The antibody of claim 1, wherein the antibody is polyclonal.

6. The antibody of claim 1, wherein the antibody is an antibody fragment.

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7. The antibody of claim 1, wherein the antibody is detectably labeled.

8. The antibody of claim 7, wherein the label is a radioisotope, a bioluminescent compound, a chemiluminescent compound, a fluorescent compound, a metal chelate, a phosphorescent compound, or an enzyme.

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9. The antibody of claim 1, wherein the antibody is bound to a carrier.

10. The antibody of claim 9, wherein the carrier is glass, polystyrene, polypropylene, polyethylene, dextran, nylon, amylase, cellulose, polyacrylamide, agarose or magnetite.

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11. The antibody of claim 1, wherein the antibody is coupled to a hapten.

12. The antibody of claim 11, wherein the hapten is biotin, dinitrophenyl, puridoxal, or fluorescein.

13. The antibody of claim 1, wherein the epitope to which the antibody binds
5 comprises at least amino acid residue 395 of SEQ ID NO: 2 or 4.

14. The antibody of claim 13, wherein the epitope to which the antibody binds comprises at least amino acid residue 395 of SEQ ID NO: 4.

10 15. The antibody of claim 1, which binds to a polypeptide comprising an amino acid sequence as set forth in SEQ ID NO: 2.

16. The antibody of claim 1, which binds to a polypeptide comprising an amino acid sequence as set forth in SEQ ID NO: 4.

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17. The antibody of claim 15, which does not bind to a polypeptide comprising an amino acid sequence as set forth in SEQ ID NO: 4.

18. The antibody of claim 16, which does not bind to a polypeptide comprising an
20 amino acid sequence as set forth in SEQ ID NO: 2

19. The antibody of claim 18, which detects an I395M mutation.

20. An antibody that binds to a mutant ABC7 polypeptide, wherein the polypeptide is
25 expressed in a subject having, or at risk for having, X-linked Sideroblastic Anemia and Ataxia.

21. The antibody of claim 20, wherein the mutant ABC7 polypeptide comprises an amino acid sequence as set forth in SEQ ID NO: 4.

22. The antibody of claim 21, wherein the antibody detects an I395M mutation in the mutant ABC7 polypeptide.

23. The antibody of claim 20, wherein the antibody is monoclonal.

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24. The antibody of claim 20, wherein the antibody is polyclonal.

25. The antibody of claim 20, wherein the antibody is an antibody fragment.

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26. A kit useful for the detection of an ATP-binding transporter-7 (ABC7) polypeptide comprising carrier means containing therein one or more containers wherein a first container contains an antibody of claim 1.

27. The kit of claim 26, wherein the antibody is detectably labeled.

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28. The kit of claim 27, wherein the label is selected from the group consisting of radioisotope, a bioluminescent compound, a chemiluminescent compound, a fluorescent compound, a metal chelate, and an enzyme.